Opis choroby *

Definicja

A rare autosomal recessive primary immunodeficiency characterized by severe reduction in the cell surface expression of HLA class I molecules, typically resulting in childhood-onset of chronic bacterial infections of the respiratory tract evolving to widespread bronchiectasis and respiratory insufficiency. Sterile necrotizing granulomatous skin lesions mainly involving the extremities and the mid-face may be observed in some patients. Severe viral infections do not occur as part of the condition. Atypical variants without respiratory or cutaneous manifestations, as well as asymptomatic individuals have been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Bare lymphocyte syndrome type 1
Bare lymphocyte syndrome type 1
MHC class I deficiency

Kod ORPHA
34592

Kod OMIM
241600

Kod ICD10
D81.6

Kod ICD11
4A01.11

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*Źródło

orphanet